Citrullinemia (CIT)

A urea cycle disorder

What is it?

Citrullinemia (also known as argininosuccinic acid synthetase deficiency (CIT)) is a urea cycle disorder. People with urea cycle disorders, like CIT, cannot properly break down and get rid of certain components of protein. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the protein, certain amino acids and a chemical called ammonia build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

What are the symptoms?

People with CIT may appear normal at birth. After a few days of life, a newborn with CIT may develop poor feeding, lack of energy, vomiting, problems breathing, or seizures. If left untreated, brain damage, coma, and death will occur. Many symptoms of CIT can be prevented by immediate treatment and lifelong management. People with CIT typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

CIT is inherited in an autosomal recessive manner. This means that for a person to be affected with CIT, he or she must have inherited two non-working copies of the gene responsible for causing CIT. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have CIT. Typically, there is no family history of CIT in an affected person. About 1 in 50,000 babies born are diagnosed with CIT.

How is it detected?

CIT may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

CIT is treated by eating a diet low in protein and drinking a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: http://www.ghr.nlm.nih.gov

Save Babies Through Screening Foundation
4 Manor View Circle
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